5(2): 28-31; 2026
Pediatric Academic Case Reports
DOI: 10.61107/pacr.2026.179

Case Report

A Rare Cause of Recurrent Pneumonia Clinic Visits: A Case Report with the Diagnosis of Severe Combined Immunodeficiency

Tekrarlayan Pnömoni Kliniğinin Nadir Bir Nedeni: Ağır Kombine İmmün Yetmezlik Tanılı Bir Olgu Sunumu

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ABSTRACT

The disease characterized by the developmental and function disorders of T and B lymphocytes together with natural killer cells is called 'Severe combined immunodeficiency'. Due to the impairment of cellular and humoral immunity, opportunistic infections such as persistent diarrhea, recurrent pneumonia attacks, chronic sinusitis and oral candidiasis begin to be seen in the first 3 months, while cases left untreated lose their lives within the first 24 months. In this study, we present a 6-month-old male child with severe combined immunodeficiency presented with pneumonia, which is among the pediatric emergencies having a high morbidity and mortal results

Keywords: *Recurrent pneumonia, Acute respiratory failure, Severe combined immunodeficiency*

ÖZET

T ve B lenfositlerin doğal öldürücü hücrelerle birlikte gelişim ve fonksiyon bozuklukları ile karakterize hastalık tablosuna 'Şiddetli kombine immün yetmezlik' adı verilir. Hücresel ve humoral immünitenin bozulması nedeniyle ilk 3 ayda persistan ishal, tekrarlayan pnömoni atakları, kronik sinüzit ve oral kandidiyazis gibi fırsatçı semptomlar görülmeye başlanırken, tedavi edilmeyen olgular ilk 24 ay içinde yaşamlarını yitirirler. Bu çalışmada, morbid ve mortal sonuçları nedeniyle pediatrik aciller arasında yer alan Şiddetli kombine immün yetmezlik tanısı konulan şiddetli pnömoni ile başvuran 6 aylık erkek hasta sunulmaktadır.

Keywords: *Tekrarlayan pnömoni, akut solunum yetmezliği, ağır kombine immün yetmezlik*

Received: 19.01.2025 · **Accepted:** 06.05.2025 · **Published:** 10.06.2026

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Cite this article as: Güzeloğlu E, Kaynar Beyaz G, Göksu AZ, Akkelle E. A rare cause of recurrent pneumonia clinic visits: a case report with the diagnosis of severe combined immunodeficiency. Pediatr Acad Case Rep. 2026;5(2):28-31.

INTRODUCTION

Severe combined immunodeficiency disease (SCID) is an inherited primary immunodeficiency disorder that presents by six months of age with opportunistic infections caused by bacteria, viruses, fungi, and protozoa. A study conducted in the USA reported that the incidence of SCID was 1 in 58,000 live births. It is one of the most serious primary immunodeficiency disorders with early death due to disturbed or absent T and B cell functions. In this study, we present a 6-month-old male patient diagnosed with Severe Combined Immunodeficiency who presented with severe pneumonia.

CASE REPORT

A 6-month-old male patient was admitted to our emergency department with fever and respiratory distress. Vital signs showed fever: 38.3 C, respiratory rate: 58/min, pulse: 162/min, BP: 80/50 mm Hg. Physical examination revealed hyperemic oropharynx, and bilateral diffuse crepitations on auscultation in the lungs. Subcostal and suprasternal retractions and nasal flaring were present. There was no hepatosplenomegaly. Laboratory tests revealed WBC (µL): 26660, Lymphocyte (µL): 2250, Hb (g/dL): 9.2, Platelet (µL): 182000, ANC (µL): 24000. Immunoglobulin values were determined as IgG: 328, IgA: 286, IgM: 56. Chest X-ray revealed consolidation areas in bilateral lower zones and diffuse ground glass appearance. In the patient's computed tomography, bilateral widespread ground-glass opacities were observed (Figure-1). It was learned from the patient's medical history that she was born at the 39th week of pregnancy by normal vaginal delivery and that she had no history of intensive care admission during the neonatal period. It was learned that she had a history of repeated hospital admissions for the last 2 months and that treatment was initiated with the diagnosis of rhinitis 2 times, otitis 1 time and pneumonia 2 time. In the patient's family history, it was stated that one of his brothers died at the age of 10 months. Since his brother did not have an epicrisis, no information could be obtained about the child's history. It could not be learned whether there was a history of immune deficiency. The patient was admitted to the ward and ceftriaxone and clarithromycin treatments were initiated. Child immunology and allergy opinions were obtained and lymphocyte subgroups were studied. As a result of lymphocyte subgroup flow cytometry analysis, the patient was diagnosed with severe combined immune deficiency and was followed up. The flow cytometry analysis results of the patient are presented in Table-1. The genetic diagnosis of the case was determined as Janus Kinase-3 (JAK-3) deficiency. Pediatric hematology was consulted during the treatment process and allogeneic hematopoietic stem cell transplantation was planned. His healthy brother was selected as the donor. Post-transplant follow-up was performed in the pediatric hematology clinic.

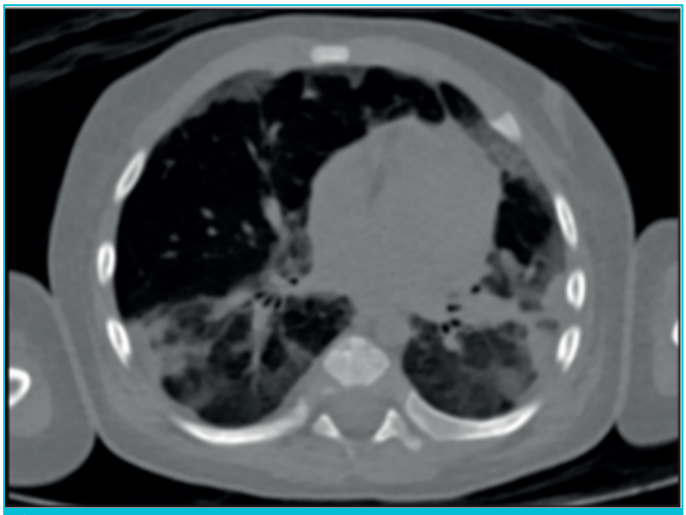

Figure 1. Extensive heterogeneous ground-glass opacities in both parenchyma

Table 1. Lymphocyte subgroups

	N	%
NK-Lymphocyte (CD3- CD16+CD56+) (µL)	4.5	0.2
T-Cytotoxic/Suppressor (CD3+CD8+) (µL)	0	0
T-Helper (CD3+CD4+) (µL)	0	0
T-Lymphocyte (CD3+) (µL)	0	0
B-Lymphocyte (CD19+) (µL)	2241	99.6
Lymphocyte (µL)	2250	8.4
White Blood Cell (WBC) (µL)	26660	100

Lymphocyte subgroup flow cytometry analysis

DISCUSSION

The Primary Immunodeficiency Treatment Consortium revised criteria for SCID (Severe combined immunodeficiency) diagnosis in 2022 to include current approaches and The 2024 Update on the classification was published by the Expert Committee of the International Union of Immunology Societies (IUIS).

Patients with typical SCID must have <0.05 × 10^9autologous T cells/L on repeat testing, pathogenic variant(s) in a SCID-associated gene, very low/undetectable T cell receptor excision loops or <20% of CD4 T cells expressing naive markers, and/or maternally engrafted T cells via the placenta. Patients with less profoundly impaired autologous T-cell differentiation are designated as having leaky/atypical SCID, with 2 or more of these: low T-cell numbers, oligoclonal T cells, low T-cell receptor excision circles, and less than 20% of CD4 T cells expressing naive markers. These patients must also have either pathogenic variant(s) in a SCID-associated gene or reduced T-cell proliferation to certain mitogens (1-5).

People with SCID have a nearly absent immune system, making them highly susceptible to recurrent, severe infections. There are several forms of SCID with varying degrees of severity, but they all share common characteristics. Newborns with SCID often have symptoms such as chronic diarrhea, thrush, skin rashes, and persistent infections that do not respond to standard treatments. Without prompt diagnosis and intervention, SCID can lead to life-threatening complications and a high risk of death. There are more than 20 possible genes involved (4-7).

Early diagnosis and appropriate treatment are essential. In this context, newborn screening is increasingly and significantly improving the prognosis of SCID. Screening for SCID in newborns is now routinely performed in many countries in Europe and the world. Data in the literature shows that survival rates for children with SCID are much higher when the disease is detected by early screening than when diagnosed later. Newborns diagnosed with SCID can receive appropriate care quickly, before serious infectious complications develop, which increases survival rates, improves quality of life, and limits side effects and treatment costs (8-11).

Treatment options for SCID primarily involve immune reconstitution, with hematopoietic stem cell transplantation (HSCT) being the best-known approach. Alternatively, gene therapy is also available for some forms of SCID. Gene therapy is an innovative treatment for Primary Immunodeficiencies (PIDs) that use autologous hematopoietic stem cell transplantation to deliver stem cells with added or edited versions of the missing or faulty gene that cause PID. Once successfully treated, SCID patients can live relatively normal lives, but may still require careful infection control measures and lifelong medical follow-up to manage potential complications. In conclusion, severe combined immunodeficiency is a rare but life-threatening genetic disorder that severely compromises the function of the immune system, making affected individuals highly vulnerable to infection (12-16).

Patient Consent Form / Hasta Onam Formu

The parents' of this patient consent was obtained for this study.

Conflict of Interest / Çıkar Çatışması

The authors declared no conflicts of interest with respect to authorship and/or publication of the article.

Financial Disclosure / Finansal Destek

The authors received no financial support for the research and/or publication of this article.

Author Contributions / Yazar Katkıları

Consept: EG,GKB; design: EG,GKB; supervison: EA; materials: EG,GKB,AZG; data collection and/or processing: EG,GKB,AZG ; analysis and interpretation: EG; literature review: EG; writing manuscript: EG; critical reviews: EA. All authors contributed to the final version of the manuscript and discussed the results and contributed to the final manuscript.

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